

Genetic Composition of Chilean Aboriginal Populations: HLA and Other Genetic Marker Variation

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ABSTRACT Phenotypes and gene frequencies for eight genetic systems are presented for five Chilean Indian tribes. Results agree with the pattern expected for Andean Indians. Genetic distances and dendrograms were obtained separately for HLA and traditional genetic markers. The similarity of both is noteworthy. Linguistic distances exhibit a correlation with genetic distances based on traditional markers. © 1996 Wiley-Liss, Inc.

Some 18 years ago Chakraborty et al. (1976) studied the genetic variation of Chilean Indians and its association with geography, language, and culture. The geographic location of the tribes was found to be the most important factor in explaining the variation in genetic distances for seven marker genes (ABO, MNSs, Rh, Diego, Duffy, Kidd, and Haptoglobin) among Indian populations.

Although extensive data on human leukocyte antigen (HLA) allele frequencies have been published worldwide, there is a lack of information regarding southern South America. The objectives of the present study are to expand the specialized literature describing HLA allele frequencies of Chilean Indians, to explore their evolutionary relationships comparing the results with those obtained by Chakraborty et al. (1976), and to collate the patterns of HLA variability with those obtained from traditional marker genes (i.e., blood groups, serum proteins, and erythrocyte enzymes) as well as with the geographic location of the tribes, linguistic differences, and cultural dissimilarities.

The Chilean Indian tribes considered in the present analysis are the following: Aymara (17° S, 70° W), Atacameño (30° S, 67° W), Pehuenche (38° S, 71° 30' W), Huilliche (40° 45' S, 73° 19' W), and Yaghan (55° 36' S, 70° 48' W). The Aymara and Ataca-

meño inhabit the desertic northern region of Chile, the Huilliche and Pehuenche the central rainforest, and the Yaghan the southern archipelagos (Fig. 1).

From the linguistic point of view, all Chilean tribes were classified by Greenberg (1960) as belonging to the Andean Ecuadorial family and the Andean subfamily, with the exception of the Atacameño, which was classified as Macro Chibchan and belonging to the Paezan subfamily. However, the Chilean Indians are quite different culturally. Their diverse patterns of subsistence, material culture, economics, and social organization are adaptations to the extremely different ecogeographic areas in which they live. The Aymara and Atacameños are small-scale agriculturalists and herders, whereas Pehuenche and Huilliche are horticulturalists and farmers, while Yaghan are nomadic canoeists (Steward, 1963).

MATERIALS AND METHODS

We studied 87 Aymara Indians, residents of Putre and Codpa, 180 Atacameño Indians living in San Pedro de Atacama, 132 Pehuenche Indians from Trapa Trapa, 105 Huilliche

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Indians from San Juan de la Costa, and 25 Yaghan from Ukika. Ten milliliters of blood was obtained from volunteers by venipuncture. Five milliliters with acid citrate dextrose (ACD) was used for determination of blood groups, erythrocyte enzymes, and plasma proteins, and 5 ml of heparinized blood was employed for HLA typing.

Antigenic specificities were determined for ABO, Rh, MNSs, Duffy, and Kidd. All determinations were carried out in tubes containing 2% red cell suspensions, using Biotest Serum Institute GMBH (Dreireich, Germany) and Gamma Biological Inc. (Houston, Texas) sera. Erythrocyte enzymes phosphoglucomutase 1 (E.C.2.7.5.1), (PGM1) and esterase D (E.C.3.1.1.1.) (EsD) and the plasma protein haptoglobin (Hp) were assessed by starch gel electrophoresis following the procedures of Spencer et al. (1964), Hopkinson et al. (1973), and Paulik (1957). HLA determinations for loci A, B, and C were performed in the field applying the micro-lymphocytotoxicity method of Terasaki and McClelland (1964). HLA antisera were obtained from Biotest Diagnostic (Dreireich, Germany) and One Lambda (Los Angeles, CA). The following specificities were included: A1, A2, A3, A10, A11, A23, A24, A28, A29, A30, A31, A32, B5, B7, B8, B13, B14, B15, B17, B18, B21, B22, B27, B35, B38, B39, B40, B41, B44, B48, Cw1, Cw2, Cw3, Cw4, Cw5, Cw6, and Cw7.

For blood groups, enzymes, and protein loci, maximum likelihood gene frequency estimators were obtained using the MAXLIK computer program of Reed and Schull (1968). Gene frequencies for the HLA system were calculated by $p = 1 - \sqrt{1 - f}$, where f is the antigen frequency (Mathiuz et al., 1970). Nei's (1978) unbiased distances were used in the generation of UPGMA (unweighted pair-group method with arithmetic averaging) dendrograms (Sneath and Sokal, 1973) employing the BIOSYS programs described in Swofford and Selander (1981).

We estimated the percentage of Indian admixture for each population according to Bernstein (1931), averaging results obtained through the use of the most informative alleles of the ABO, Rh, MNSs, Duffy, and haptoglobin systems. The gene frequencies of

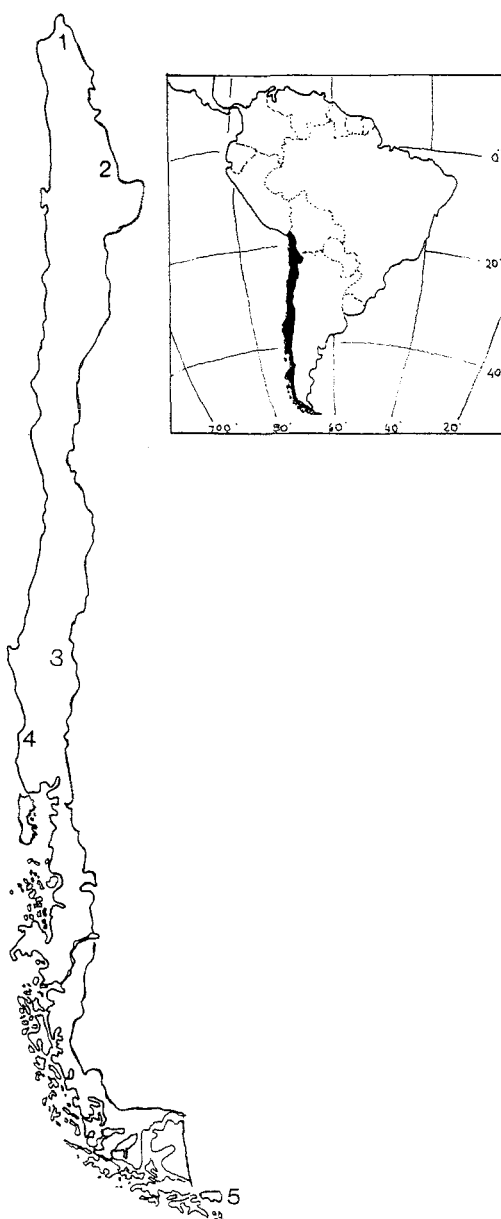


Fig. 1. Map of Chile showing the geographic location of the five Chilean Indian tribes included in the present study. 1, Aymara; 2, Atacameño; 3, Pehuenche; 4, Huilliche; 5, Yaghan.

the ancestral populations (Spaniards and Mapuche Indians) were obtained from Matson et al. (1967), Ferrell et al. (1978), Nagel and Echeverry (1963), Campillo (1976),

Guasch (1950), and Agosti Romero et al. (1950).

Linguistic distances and cultural dissimilarity indices were computed according to the methods described in Chakraborty et al. (1976). Briefly, the linguistic classification of South American Indians as given by Greenberg (1960) consists of a nested hierarchy of family, subfamily, and group. Although this classification does not take into account all the dissimilarities between these five tribal languages, alternatives do not offer substantial advantages.

Linguistic distance is taken as zero if the languages belong to the same group; the unit distance is ascribed to languages belonging to two different groups of the same subfamily; and a distance of three is assigned between languages belonging to two different families. In principle, a distance of two units would correspond to languages belonging to two different subfamilies of the same family. However, such a situation does not arise in these five languages according to Greenberg's classification for a similar analysis of linguistic diversity (see Llop and Rothhammer, 1974).

The cultural attributes were taken from the comparative study of Driver and Massey (1957), and the broad classification was drawn from the descriptions given in the *Handbook of South American Indians*, Vols. 1 and 2 (Steward, 1963). The cultural variables were subsistence (type of subsistence, hunting, fishing, farming, food preparation), material culture (transportation, housing and architecture, clothing, weapons), economics (division of labor, property, and inheritance) and social organization (marriage, family, kinship, and groups). Numeric values were assigned to each attribute representing a gradation in the degree of acculturation. Absolute differences of these numeric values for each of the attributes were computed, and the sum represents the total cultural dissimilarities between the two tribes concerned. The values were then standardized by dividing each of them by the maximum (Chakraborty et al., 1976).

Pairwise correlations among genetic, geographic, linguistic, and cultural distances were computed using Mantel's (1967) test of matrix correspondence, with the extensions

proposed by Smouse et al. (1986), employing a computer program developed by J. Long.

RESULTS

The phenotype and allele frequency distributions for eight marker systems in five Chilean Indian tribes are presented in Tables 1 and 2. It is noteworthy that gene frequencies are consistent with the patterns expected for Andean Indians (i.e., high frequencies of ABO*O, Fy*a, HLA*A2, HLA*A24, HLA*A28, HLA*B35, HLA*B39, and HLA*B40 alleles as well as Rh*DcE, Rh*DcE, and L*Ms haplotypes).

The analysis of admixture in the studied populations indicates that the Pehuenche and Aymara exhibit the lowest mean contribution of nonaboriginal genes (Spanish) to their genomes (i.e., 0.05 ± 0.02 and 0.08 ± 0.01). The Atacameño and Huilliche exhibit, respectively, 0.09 ± 0.05 and 0.15 ± 0.11 mean admixture. The Yaghan are the most admixed group, with a mean proportion of foreign genes of 0.26 ± 0.08 .

Table 3 shows genetic distances among tribes for HLA genes (upper triangle) and for the remaining genetic markers studied (lower triangle). For the HLA system, the smallest genetic distances were observed between Pehuenche and Huilliche and Huilliche and Yaghan. For blood groups, enzymes, and proteins, the smallest genetic distances were obtained between Pehuenche and Huilliche and Pehuenche and Yaghan.

Figure 2 shows two dendrograms based on HLA genes and on blood groups, serum proteins, and enzymes. The good agreement of both is evident.

DISCUSSION

The present study increases our knowledge of HLA and marker gene data for aboriginal groups of southern South America and at the same time clarifies the relationships among Chilean aboriginal populations. Our dendrograms, based on a larger number of marker loci, produced more consistent results than Chakraborty et al.'s (1976) analysis. The Aymara and Atacameño, both Andean agriculturalists, cluster together (Fig. 2), as well as the Pehuenche and Huilliche, who are southern horticulturalists and farmers. The second cluster also includes the

TABLE 1. Phenotype distribution in five Chilean Indian tribes¹

System	Phenotypes	Observed number of individuals in each class				
		Aymara	Atacameño	Pehuenche	Huilliche	Yaghan
ABO	A	5	20	7	10	2
	B	2	1	4	7	2
	O	80	161	124	87	20
Total		87	182	135	104	24
	DCE	1	0	0	0	0
	DCEe	1	6	2	1	0
	DCe	13	36	59	61	6
	DCcE	14	10	2	0	0
	DCcEe	28	83	51	31	7
	DCce	2	11	3	5	4
	DcE	23	30	16	4	3
	DcEe	5	5	2	3	4
Total		87	181	135	105	24
	MS	6	10	4	0	3
	MSs	9	24	13	7	4
	Ms	18	49	45	49	6
	MNS	3	6	2	2	0
	MNSs	16	20	12	5	5
	MNs	15	48	49	32	6
	NS	2	0	0	0	0
	NSs	6	4	2	0	0
	Ns	11	18	8	0	0
Total		86	179	135	95	24
Duffy	a	65	113	73	57	14
	ab	15	48	49	37	8
	b	2	19	13	10	2
Total		82	180	135	104	24
Kidd	a	7	—	1	8	3
	ab	34	—	39	27	10
	a	39	—	95	70	11
Total		80		135	105	24
Phosphoglucos mutase	1-1	51	138	36	55	10
	2-1	31	32	65	31	11
	2-2	5	2	30	18	3
Total		87	172	131	104	24
Esterase D	1-1	53	65	89	85	20
	2-1	30	83	24	16	4
	2-2	4	25	4	3	0
Total		87	173	117	104	24
Haptoglobin	1-1	38	80	72	49	6
	2-1	39	85	50	36	12
	2-2	10	10	10	19	4
Total		87	175	132	104	22
HLA	A1	3	2	1	4	0
	A2	68	21	27	45	13
	A3	0	1	0	10	2
	A10	1	0	1	4	0
	A11	1	0	0	3	1
	A23	0	0	0	0	1
	A24	22	5	15	25	5
	A28	11	2	37	62	9
	A29	0	0	0	5	1
	A30	0	0	0	1	0
	A31	9	22	16	12	1
	A32	0	0	0	0	0

continued

TABLE 1. *Continued*

System	Phenotypes	Observed number of individuals in each class				
		Aymara	Atacameño	Pehuenche	Huilliche	Yaghan
HLA	B5	10	1	7	14	2
	B7	11	0	0	5	2
	B8	0	0	0	6	0
	B13	1	1	0	3	0
	B14	0	6	0	6	2
	B15	8	17	11	11	5
	B17	0	0	0	6	0
	B18	1	0	0	1	0
	B21	0	0	0	3	4
	B22	0	0	0	0	0
	B27	0	0	0	1	0
	B35	54	9	15	20	0
	B38	0	0	0	2	0
	B39	6	1	35	60	1
	B40	24	10	8	16	10
	B41	—	—	—	1	0
	B44	1	0	0	8	2
	B48	—	—	—	2	0
HLA	CW1	13	14	14	7	2
	CW2	0	4	1	0	1
	CW3	15	0	10	20	11
	CW4	51	8	13	24	5
	CW5	1	0	1	4	1
	CW6	—	—	0	1	0
	CW7	—	—	8	72	3
Total		78	30	56	98	19

¹ A dash indicates that the test has not been performed.

Yaghan, nomadic canoeists of the extreme south, indicating a microevolutionary relationship among southern tribal population. In this respect the genetic data is in agreement with archaeological information and craniometrical studies performed in Chilean prehistoric populations (Rothhammer et al., 1986).

The correlation of genetic distances with language is satisfactory despite the fact that the Atacameño are linguistically divergent from the other groups according to Greenberg (1960) and that linguistic classifications are static and do not reflect past microevolutionary events occurring in populations.

In general, empirical evidence has shown that multilocus approaches are needed to yield more consistent patterns of relationships among populations. Our results indicate that three loci of the major human histocompatibility complex yield essentially the same results as a groups of non-HLA protein markers (Table 4). This fact alone is noteworthy, but also because it is generally assumed that selective forces are acting on the HLA

system, making this system a bad choice for population comparisons.

The high number of HLA alleles probably compensate for the low number of loci and mask the effect of selective forces acting on single alleles.

The genetic distances for HLA are on average larger than corresponding distances for traditional markers, thus reflecting the higher levels of variability encountered for the first group of markers.

Finally, as expected, geographic and cultural distances are strongly correlated, given the fact that cultural dissimilarities represent different adaptations to contrasting eco-geographic areas.

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TABLE 2. Gene frequencies in five Chilean Indian tribes¹

System	Allele or haplotype	Gene frequencies				
		Aymara	Atacameño	Pehuenche	Huilliche	Yaghan
ABO	ABO*A	0.029	0.056	0.026	0.054	0.043
	ABO*B	0.012	0.003	0.015	0.039	0.043
	ABO*O	0.959	0.941	0.959	0.907	0.914
Rh	Rh*DCE	0.102	0.046	0.015	0.005	0.000
	Rh*Dce	0.323	0.473	0.644	0.757	0.479
	Rh*DcE	0.530	0.434	0.322	0.200	0.354
	Rh*Dce	0.013	0.027	0.006	0.110	0.048
	Rh*dce	0.032	0.020	0.014	0.027	0.119
	L*MS	0.192	0.178	0.113	0.056	0.313
MNSs	L*Ms	0.390	0.492	0.579	0.739	0.458
	L*NS	0.116	0.045	0.031	0.028	0.000
	L*Ns	0.302	0.285	0.276	0.177	0.229
Duffy	Fy*a	0.884	0.742	0.722	0.726	0.750
	Fy*b	0.116	0.258	0.278	0.274	0.250
Kidd	JK*a	0.300	—	0.152	0.205	0.333
	JK*b	0.700	—	0.848	0.795	0.666
Phosphogluco mutase	PGM*1	0.764	0.906	0.5.23	0.678	0.646
	PGM*2	0.236	0.094	0.477	0.322	0.354
Esterase D	EsD*1	0.782	0.616	0.863	0.894	0.917
	EsD*2	0.218	0.384	0.137	0.106	0.083
Haptoglobin	Hp*1	0.661	0.700	0.735	0.644	0.545
	Hp*2	0.339	0.300	0.265	0.356	0.454
HLA	HLA*A1	0.019	0.340	0.008	0.021	0.000
	HLA*A2	0.638	0.423	0.280	0.265	0.430
	HLA*A3	0.000	0.017	0.000	0.052	0.050
	HLA*A10	0.060	0.000	0.000	0.021	0.000
	HLA*A11	0.000	0.000	0.000	0.015	0.026
	HLA*A23	0.000	0.000	0.000	0.000	0.026
	HLA*A24	0.151	0.087	0.143	0.137	0.141
	HLA*A28	0.073	0.034	0.415	0.393	0.275
	HLA*A29	0.000	0.000	0.000	0.026	0.026
	HLA*A30	0.000	0.000	0.000	0.005	0.000
	HLA*A31	0.059	0.394	0.153	0.063	0.027
	Blank	0.000	0.011	0.000	0.002	0.000
	HLA*B5	0.066	0.017	0.065	0.075	0.054
	HLA*B7	0.073	0.000	0.000	0.026	0.054
	HLA*B8	0.000	0.000	0.000	0.032	0.000
	HLA*B13	0.006	0.017	0.000	0.016	0.000
	HLA*B14	0.000	0.106	0.008	0.032	0.054
	HLA*B15	0.053	0.340	0.104	0.059	0.142
	HLA*B17	0.000	0.000	0.000	0.032	0.000
	HLA*B18	0.006	0.000	0.000	0.005	0.000
	HLA*B21	0.000	0.000	0.000	0.015	0.112
	HLA*B27	0.000	0.000	0.000	0.005	0.000
	HLA*B35	0.446	0.163	0.144	0.107	0.000
	HLA*B38	0.000	0.000	0.000	0.010	0.000
	HLA*B39	0.039	0.170	0.387	0.378	0.027
	HLA*B40	0.168	0.184	0.075	0.085	0.311
	HLA*B41	—	—	—	0.005	0.000
	HLA*B44	0.006	0.000	0.000	0.042	0.054
	HLA*B48	—	—	—	0.010	0.000
	Blank	0.137	0.156	0.217	0.081	0.192
	HLA*Cw1	0.085	0.270	0.134	0.036	0.054
	HLA*CW2	0.000	0.069	0.008	0.000	0.027
	HLA*CW3	0.131	0.000	0.094	0.108	0.350
	HLA*CW4	0.401	0.144	0.124	0.131	0.142
	HLA*CW5	0.006	0.000	0.008	0.021	0.027
	HLA*CW6	—	—	0.000	0.005	0.000
	HLA*CW7	—	—	0.075	0.485	0.082
	Blank	0.377	0.517	0.557	0.214	0.318

¹The alleles absent in all the studied populations are not included in the table. A dash indicates that the test has not been performed.

TABLE 3. Nei's standard genetic distance matrix for HLA (upper triangle) and seven other genetic markers (lower half triangle) on five Chilean Indian tribes

	Aymara	Atacameño	Pehuenche	Huilliche	Yaghan
Aymara	—	0.2637	0.3679	0.4025	0.2377
Atacameño	0.0197	—	0.2722	0.3741	0.2786
Pehuenche	0.0443	0.0549	—	0.0162	0.2171
Huilliche	0.0627	0.0594	0.0128	—	0.2261
Yaghan	0.0198	0.0417	0.0161	0.020	—

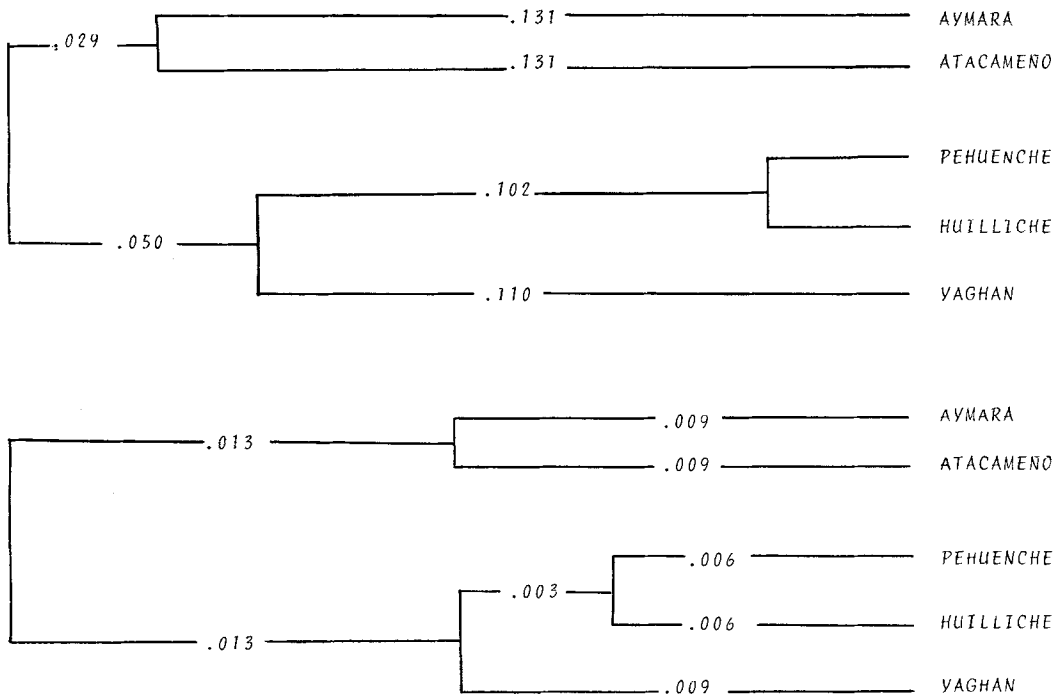


Fig. 2. Dendrograms obtained for five Chilean Indian tribes, considering 33 alleles for the HLA system (top) and 22 alleles for the other seven genetic systems (bottom).

TABLE 4. Mantel correlation coefficients of genetic geographic, and cultural distances in five Chilean tribes

	Genetic distance 2 ¹	Linguistic distance	Geographic distance	Cultural dissimilarity index
Genetic distance 1 ²	0.78**	0.39	0.47	0.36
Genetic distance 2 ¹	—	0.50*	0.35	0.03
Linguistic distance	—	—	0.10	0.06
Geographic distance	—	—	—	0.87***

¹ Genetic distance for blood groups, proteins, and enzymes.² Genetic distance for HLA only.* $P < 0.05$.** $P < 0.01$.*** $P < 0.001$.

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